CDT1 gene

chromatin licensing and DNA replication factor 1

Normal Function

The *CDT1* gene provides instructions for making a protein that is important in the copying of a cell's DNA before the cell divides (a process known as DNA replication). The protein produced from this gene is one of a group of proteins known as the pre-replication complex. In a multi-step process, the components of this complex attach (bind) to certain regions of DNA known as origins of replication (or origins), where the process of DNA copying begins. When the pre-replication complex is attached to the origin, replication is able to begin at that location. This tightly controlled process, called replication licensing, helps ensure that DNA replication occurs only once per cell division and is required for cells to divide.

Health Conditions Related to Genetic Changes

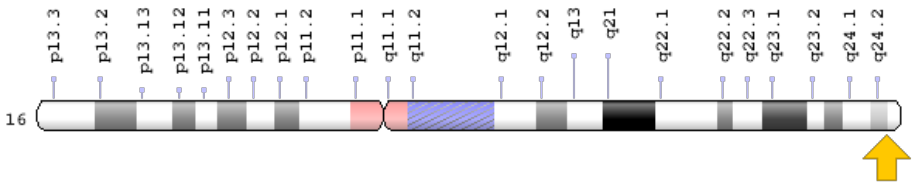
Meier-Gorlin syndrome

Mutations in the *CDT1* gene cause Meier-Gorlin syndrome, a condition characterized by short stature, underdeveloped kneecaps, and small ears. These mutations alter the CDT1 protein, typically by changing single protein building blocks (amino acids) or by leading to production of an abnormally short version of the CDT1 protein. As a result, assembly of the pre-replication complex is impaired, which disrupts replication licensing; however, it is not clear how a reduction in replication licensing leads to Meier-Gorlin syndrome. Researchers speculate that such a reduction delays the cell division process, which slows growth of the bones and other tissues during development. It is not known why development of the kneecaps and ears is particularly affected.

Chromosomal Location

Cytogenetic Location: 16q24.3, which is the long (q) arm of chromosome 16 at position 24.3

Molecular Location: base pairs 88,803,778 to 88,809,258 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CDT1_HUMAN
- DNA replication factor Cdt1
- Double parked, Drosophila, homolog of
- DUP
- RIS2

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): DNA Synthesis Begins at Replication Origins
  https://www.ncbi.nlm.nih.gov/books/NBK26826/#_A796_
- The Cell: A Molecular Approach (second edition, 2000): Origins and the Initiation of Replication
  https://www.ncbi.nlm.nih.gov/books/NBK9940/#_A789_

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CDT1%5BTIAB%5D%29+OR+%28chromatin+licensing+and+DNA+replication+factor+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1080+days%22%5Bdp%5D

OMIM

- CHROMATIN LICENSING AND DNA REPLICATION FACTOR 1
  http://omim.org/entry/605525

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/CDT1ID44175ch16q24.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CDT1%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=24576
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/81620
- UniProt
  http://www.uniprot.org/uniprot/Q9H211

Sources for This Summary

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